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Asymmetrical Coronal Synostosis, Cutaneous Syndactyly of the Fingers and Toes, and Jejunal Atresia in a Male Infant

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We report on a male infant with an unusual type of acrocephalosyndactyly presenting with unilateral coronal craniosynostosis, cutaneous syndactyly of toes 2 and 3, loss of distal triradii, and transverse alignment of dermal ridges of the palm suggesting syndactyly, atresia of the proximal jejunum, and anal stenosis. © 1996 Wiley-Liss, Inc.

KEY WORDS: multiple malformations, acrocephalosyndactyly, jejunum atresia

INTRODUCTION

Craniosynostosis is the heuristic hallmark of a nosological class of heterogeneous malformation syndromes with a special predilection of abnormalities of hands and feet. Apart from well known types, the molecular origin of which is being explored, single entities, mostly sporadic, remain to be defined. Published observations of this type have been reported, reviewed, and analysed systematically by Cohen [1986]. This list is being continued by the case here reported which is unusual because of the association with intestinal atresia.

CLINICAL REPORT

MW 930605 is the first child of healthy non-consanguineous parents. At his birth both parents were 28 years old. Pregnancy was normal. The amount of amniotic fluid was said to be normal. Delivery by cesarean section was performed 6 weeks before term because of the spontaneous rupture of the amniotic membranes. Weight was 2,630 g, length was 45 cm, and OFC was 32.5 cm.

An asymmetrical deformity of the skull was noted at birth. The left orbit was laterally displaced and the globe was protruding. Stenosis of the upper intestinal tract which had been suspected by antenatal ultrasonography was shown to be due to atresia of the proximal jejunum and resection was performed at the 2nd day. Postoperative course was uneventful. Histologic studies of the resected bowel did not show aganglionosis. Additional anal stenosis was treated conservatively. No other abnormalities were discovered.

Radiography and cranial computerized tomography supported the diagnosis of unilateral coronal synostosis. Morphology of the brain appeared normal. Neither optic atrophy nor any abnormality of the eyes was observed. The fused coronary suture was excised at the age of 11 months. Craniofacial deformation has improved continuously since then.

Psychomotor development has been normal, but mild asymmetric muscular hypertonia persists. At age 2½ years height was 91 cm, weight was 12.5 kg, and OFC was 48 cm (all measurements near the 50th centile).

The 2nd and 3rd toes were completely fused. No cutaneous syndactyly of the fingers was present but dermatoglyphics were unusual in that the direction of distal palmar ridges was transverse and b and d triradii were absent. The digital patterns consisted of ulnar loops only (Fig. 1). Palmar creases also were transverse. Radiographs of the hands and of the feet at age 2 years showed that bone age and all skeletal parts were normal for age.

Dermatoglyphics of the parents showed normal digital and palmar patterns and position of the distal palmar triradii (Fig. 1). Palmar creases were normal.

DISCUSSION

The present observation falls into the class of unusual acrocephalosyndactylies (ACS) because of premature asymmetrical fusion of the coronal suture and cutaneous syndactyly of toes. Although there was no visible syndactyly of the fingers palmar dermatoglyphics were similar to those noted in syndactyly.

The study of dermatoglyphics in various degrees of syndactyly showed that interdigital cutaneous fusions

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Dedicated to Jürgen W. Spranger on the occasion of his 65th birthday with admiration and best wishes.

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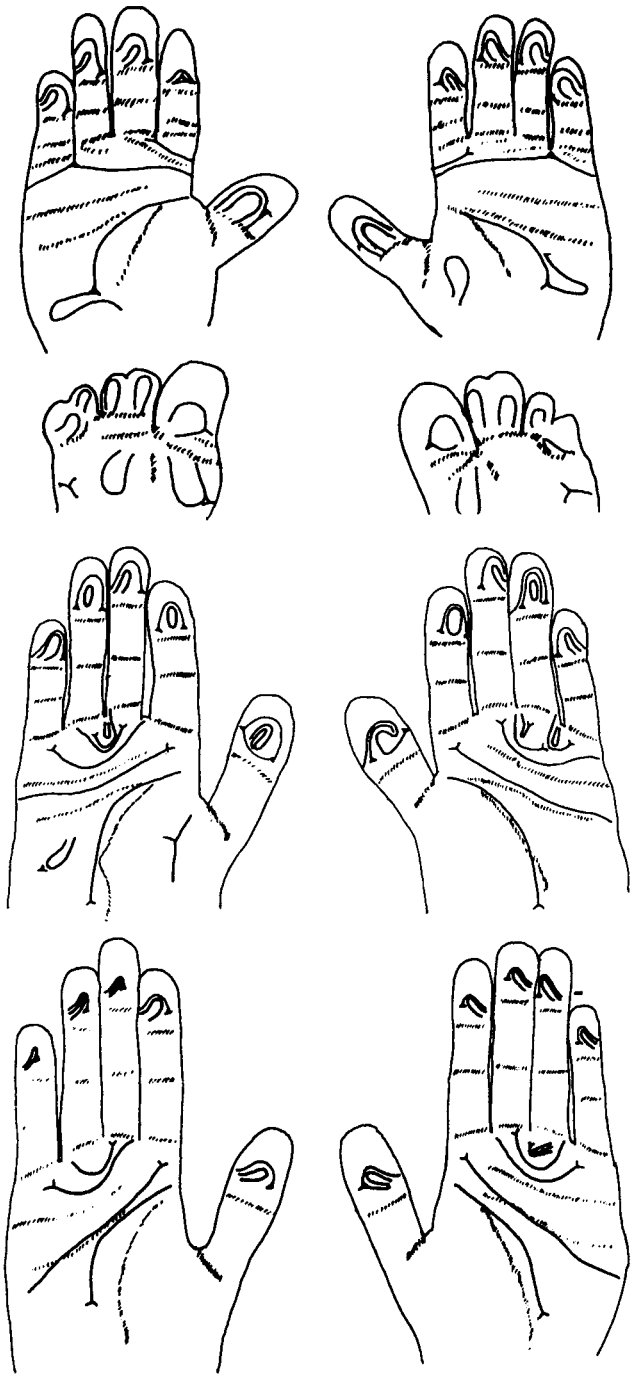

Fig. 1. Palmar and plantar dermatoglyphics of the patient and his parents.

present with approximation, fusion and hence loss of digital triradii and transversely aligned ridges [Schaumann and Alter, 1976]. Indeed in patients with ACS type III (Saethre-Chotzen) a low a-b ridge count was considered an indication of syndactyly between digits 2 and 3 although no actual syndactyly had been found [Schaumann and Pantke, 1978]. However this tendency may be masked by more complex dermatoglyphic patterns in patients with ACS I (Apert) with multiple syndactylies [Scholz, 1967]. In the study of ACS III [Schaumann and Alter, 1976] the distribution of digital patterns of the patients differed from controls but not from relatives. Here the uniform pattern of digital loops can be related to the parental patterns which consist of loops and whorls only.

The patient does not bear resemblance to any of the known types of ACS or published single cases summarized by Cohen [1986]. Therefore this association in one case could be coincidental. In a study of visceral anomalies in Apert syndrome [Cohen and Kreiborg, 1993], no such malformation was noted; anomalies of the gut were not noted in reviews of patients with coronal craniosynostosis [Anderson and Paranhos Gomes, 1968; Shillitoe and Matson, 1968; Hunter and Rudd, 1977]. A possibly "new" MCA/MR syndrome with ACS [Pfeiffer et al., 1987] shares some manifestations but premature craniosynostosis was sagittal. It is noteworthy that jejunal atresia was noticed in a case of scaphocephaly [Anderson and Paranhos Gomes, 1968].

As aganglionosis was excluded this observation cannot be compared with various craniofacial syndromes which present with Hirschsprung disease [Yomo et al., 1991; Fryer, 1993].

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